

SEQUENCE LISTING.txt

SEQUENCE LISTING

<110> Omary, Bishr
Ku, Nam-Om

<120> Keratin 8 and 18 mutations are Risk
Factors for Developing Liver Disease of Multiple Etiologies

<130> STAN-297WO

<140> 10/552,949

<141> 2006-07-27

<150> 60/462,989

<151> 2003-04-14

<160> 9

<170> FastSEQ for windows Version 4.0

<210> 1

<211> 1485

<212> DNA

<213> H. sapiens

<220>

<221> CDS

<222> (119)...(1408)

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agc ttc acc act cgc tcc acc ttc tcc acc aac tac cgg tcc ctg ggc						166
Ser Phe Thr Thr Arg Ser Thr Phe Ser Thr Asn Tyr Arg Ser Leu Gly						
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tct gtc cag gcg ccc agc tac ggc gcc cgg ccg gtc agc agc gcg gcc	214
Ser Val Gln Ala Pro Ser Tyr Gly Ala Arg Pro Val Ser Ser Ala Ala	
	20 25 30

agc gtc tat gca ggc gct ggg ggc tct ggt tcc cgg atc tcc gtg tcc	262
Ser Val Tyr Ala Gly Ala Gly Gly Ser Gly Ser Arg Ile Ser Val Ser	
	35 40 45

cgc tcc acc agc ttc agg ggc ggc atg ggg tcc ggg ggc ctg gcc acc	310
Arg Ser Thr Ser Phe Arg Gly Gly Met Gly Ser Gly Gly Leu Ala Thr	
	50 55 60

ggg ata gcc ggg ggt ctg gca gga atg gga ggc atc cag aac gag aag	358
Gly Ile Ala Gly Gly Leu Ala Gly Met Gly Gly Ile Gln Asn Glu Lys	
	65 70 75 80

gag acc atg caa agc ctg aac gac cgc ctg gcc tct tac ctg gac aga	406
Glu Thr Met Gln Ser Leu Asn Asp Arg Leu Ala Ser Tyr Leu Asp Arg	
	85 90 95

gtg agg agc ctg gag acc gag aac cgg agg ctg gag agc aaa atc cgg	454
Val Arg Ser Leu Glu Thr Glu Asn Arg Arg Leu Glu Ser Lys Ile Arg	
	100 105 110

gag cac ttg gag aag aag gga ccc cag gtc aga gac tgg agc cat tac	502
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SEQUENCE LISTING.txt																
Glu	His	Leu	Glu	Lys	Lys	Gly	Pro	Gln	Val	Arg	Asp	Trp	Ser	His	Tyr	
		115					120					125				
ttc	aag	atc	atc	gag	gac	ctg	agg	gct	cag	atc	ttc	gca	aat	act	gtg	550
Phe	Lys	Ile	Ile	Glu	Asp	Leu	Arg	Ala	Gln	Ile	Phe	Ala	Asn	Thr	Val	
	130					135					140					
gac	aat	gcc	cgc	atc	gtt	ctg	cag	att	gac	aat	gcc	cgt	ctt	gct	gct	598
Asp	Asn	Ala	Arg	Ile	Val	Leu	Gln	Ile	Asp	Asn	Ala	Arg	Leu	Ala	Ala	
145					150					155					160	
gat	gac	ttt	aga	gtc	aag	tat	gag	aca	gag	ctg	gcc	atg	cgc	cag	tct	646
Asp	Asp	Phe	Arg	Val	Lys	Tyr	Glu	Thr	Glu	Leu	Ala	Met	Arg	Gln	Ser	
				165					170					175		
gtg	gag	aac	gac	atc	cat	ggg	ctc	cgc	aag	gtc	att	gat	gac	acc	aat	694
Val	Glu	Asn	Asp	Ile	His	Gly	Leu	Arg	Lys	Val	Ile	Asp	Asp	Thr	Asn	
			180					185					190			
atc	aca	cga	ctg	cag	ctg	gag	aca	gag	atc	gag	gct	ctc	aag	gag	gag	742
Ile	Thr	Arg	Leu	Gln	Leu	Glu	Thr	Glu	Ile	Glu	Ala	Leu	Lys	Glu	Glu	
		195					200					205				
ctg	ctc	ttc	atg	aag	aag	aac	cac	gaa	gag	gaa	gta	aaa	ggc	cta	caa	790
Leu	Leu	Phe	Met	Lys	Lys	Asn	His	Glu	Glu	Glu	Val	Lys	Gly	Leu	Gln	
		210				215					220					
gcc	cag	att	gcc	agc	tct	ggg	ttg	acc	gtg	gag	gta	gat	gcc	ccc	aaa	838
Ala	Gln	Ile	Ala	Ser	Ser	Gly	Leu	Thr	Val	Glu	Val	Asp	Ala	Pro	Lys	
225					230					235					240	
tct	cag	gac	ctc	gcc	aag	atc	atg	gca	gac	atc	cgg	gcc	caa	tat	gac	886
Ser	Gln	Asp	Leu	Ala	Lys	Ile	Met	Ala	Asp	Ile	Arg	Ala	Gln	Tyr	Asp	
				245					250					255		
gag	ctg	gct	cgg	aag	aac	cga	gag	gag	cta	gac	aag	tac	tgg	tct	cag	934
Glu	Leu	Ala	Arg	Lys	Asn	Arg	Glu	Glu	Leu	Asp	Lys	Tyr	Trp	Ser	Gln	
			260					265					270			
cag	att	gag	gag	agc	acc	aca	gtg	gtc	acc	aca	cag	tct	gct	gag	gtt	982
Gln	Ile	Glu	Glu	Ser	Thr	Thr	Val	Val	Thr	Thr	Gln	Ser	Ala	Glu	Val	
		275					280					285				
gga	gct	gct	gag	acg	acg	ctc	aca	gag	ctg	aga	cgt	aca	gtc	cag	tcc	1030
Gly	Ala	Ala	Glu	Thr	Thr	Leu	Thr	Glu	Leu	Arg	Arg	Thr	Val	Gln	Ser	
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ttg	gag	atc	gac	ctg	gac	tcc	atg	aga	aat	ctg	aag	gcc	agc	ttg	gag	1078
Leu	Glu	Ile	Asp	Leu	Asp	Ser	Met	Arg	Asn	Leu	Lys	Ala	Ser	Leu	Glu	
305					310					315					320	
aac	agc	ctg	agg	gag	gtg	gag	gcc	cgc	tac	gcc	cta	cag	atg	gag	cag	1126
Asn	Ser	Leu	Arg	Glu	Val	Glu	Ala	Arg	Tyr	Ala	Leu	Gln	Met	Glu	Gln	
				325					330					335		
ctc	aac	ggg	atc	ctg	ctg	cac	ctt	gag	tca	gag	ctg	gca	cag	acc	cgg	1174
Leu	Asn	Gly	Ile	Leu	Leu	His	Leu	Glu	Ser	Glu	Leu	Ala	Gln	Thr	Arg	
			340					345					350			
gca	gag	gga	cag	cgc	cag	gcc	cag	gag	tat	gag	gcc	ctg	ctg	aac	atc	1222
Ala	Glu	Gly	Gln	Arg	Gln	Ala	Gln	Glu	Tyr	Glu	Ala	Leu	Leu	Asn	Ile	
		355					360					365				

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aag gtc aag ctg gag gct gag atc gcc acc tac cgc cgc ctg ctg gaa	1270
Lys Val Lys Leu Glu Ala Glu Ile Ala Thr Tyr Arg Arg Leu Leu Glu	
370 375 380	
gat ggc gag gac ttt aat ctt ggt gat gcc ttg gac agc agc aac tcc	1318
Asp Gly Glu Asp Phe Asn Leu Gly Asp Ala Leu Asp Ser Ser Asn Ser	
385 390 395 400	
atg caa acc atc caa aag acc acc acc cgc cgg ata gtg gat ggc aaa	1366
Met Gln Thr Ile Gln Lys Thr Thr Thr Arg Arg Ile Val Asp Gly Lys	
405 410 415	
gtg gtg tct gag acc aat gac acc aaa gtt ctg agg cat taa	1408
Val Val Ser Glu Thr Asn Asp Thr Lys Val Leu Arg His *	
420 425	
gccagcagaa gcagggtacc ctttggggag caggaggcca ataaaaagtt cagagttcaa	1468
aaaaaaaaaa aaaaaaa	1485

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 <213> H. sapiens

<400> 2

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35 40 45	
Arg Ser Thr Ser Phe Arg Gly Gly Met Gly Ser Gly Gly Leu Ala Thr	
50 55 60	
Gly Ile Ala Gly Gly Leu Ala Gly Met Gly Gly Ile Gln Asn Glu Lys	
65 70 75 80	
Glu Thr Met Gln Ser Leu Asn Asp Arg Leu Ala Ser Tyr Leu Asp Arg	
85 90 95	
Val Arg Ser Leu Glu Thr Glu Asn Arg Arg Leu Glu Ser Lys Ile Arg	
100 105 110	
Glu His Leu Glu Lys Lys Gly Pro Gln Val Arg Asp Trp Ser His Tyr	
115 120 125	
Phe Lys Ile Ile Glu Asp Leu Arg Ala Gln Ile Phe Ala Asn Thr Val	
130 135 140	
Asp Asn Ala Arg Ile Val Leu Gln Ile Asp Asn Ala Arg Leu Ala Ala	
145 150 155 160	
Asp Asp Phe Arg Val Lys Tyr Glu Thr Glu Leu Ala Met Arg Gln Ser	
165 170 175	
Val Glu Asn Asp Ile His Gly Leu Arg Lys Val Ile Asp Asp Thr Asn	
180 185 190	
Ile Thr Arg Leu Gln Leu Glu Thr Glu Ile Glu Ala Leu Lys Glu Glu	
195 200 205	
Leu Leu Phe Met Lys Lys Asn His Glu Glu Glu Val Lys Gly Leu Gln	
210 215 220	
Ala Gln Ile Ala Ser Ser Gly Leu Thr Val Glu Val Asp Ala Pro Lys	
225 230 235 240	
Ser Gln Asp Leu Ala Lys Ile Met Ala Asp Ile Arg Ala Gln Tyr Asp	
245 250 255	
Glu Leu Ala Arg Lys Asn Arg Glu Glu Leu Asp Lys Tyr Trp Ser Gln	
260 265 270	
Gln Ile Glu Glu Ser Thr Thr Val Val Thr Thr Gln Ser Ala Glu Val	
275 280 285	
Gly Ala Glu Thr Thr Leu Thr Glu Leu Arg Arg Thr Val Gln Ser	
290 295 300	

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Leu Glu Ile Asp Leu Asp Ser Met Arg Asn Leu Lys Ala Ser Leu Glu
 305 310 315 320
 Asn Ser Leu Arg Glu Val Glu Ala Arg Tyr Ala Leu Gln Met Glu Gln
 325 330 335
 Leu Asn Gly Ile Leu Leu His Leu Glu Ser Glu Leu Ala Gln Thr Arg
 340 345 350
 Ala Glu Gly Gln Arg Gln Ala Gln Glu Tyr Glu Ala Leu Leu Asn Ile
 355 360 365
 Lys Val Lys Leu Glu Ala Glu Ile Ala Thr Tyr Arg Arg Leu Leu Glu
 370 375 380
 Asp Gly Glu Asp Phe Asn Leu Gly Asp Ala Leu Asp Ser Ser Asn Ser
 385 390 395 400
 Met Gln Thr Ile Gln Lys Thr Thr Thr Arg Arg Ile Val Asp Gly Lys
 405 410 415
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<220>
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 <222> (63)...(1511)

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 Ser Ile Arg Val Thr Gln Lys Ser Tyr Lys Val Ser Thr Ser Gly 15
 1 5 10 15
 ccc cgg gcc ttc agc agc cgc tcc tac acg agt ggg ccc ggt tcc cgc 155
 Pro Arg Ala Phe Ser Ser Arg Ser Tyr Thr Ser Gly Pro Gly Ser Arg 20 25 30
 atc agc tcc tcg agc ttc tcc cga gtg ggc agc agc aac ttt cgc ggt 203
 Ile Ser Ser Ser Ser Phe Ser Arg Val Gly Ser Ser Asn Phe Arg Gly 35 40 45
 ggc ctg ggc ggc ggc tat ggt ggg gcc agc ggc atg gga ggc atc acc 251
 Gly Leu Gly Gly Gly Tyr Gly Gly Ala Ser Gly Met Gly Gly Ile Thr 50 55 60
 gca gtt acg gtc aac cag agc ctg ctg agc ccc ctt gtc ctg gag gtg 299
 Ala Val Thr Val Asn Gln Ser Leu Leu Ser Pro Leu Val Leu Glu Val 65 70 75
 gac ccc aac atc cag gcc gtg cgc acc cag gag aag gag cag atc aag 347
 Asp Pro Asn Ile Gln Ala Val Arg Thr Gln Glu Lys Glu Gln Ile Lys 80 85 90 95
 acc ctc aac aac aag ttt gcc tcc ttc ata gac aag gta cgg ttc ctg 395
 Thr Leu Asn Asn Lys Phe Ala Ser Phe Ile Asp Lys Val Arg Phe Leu 100 105 110
 gag cag cag aac aag atg ctg gag acc aag tgg agc ctc ctg cag cag 443
 Glu Gln Gln Asn Lys Met Leu Glu Thr Lys Trp Ser Leu Leu Gln Gln 115 120 125
 cag aag acg gct cga agc aac atg gac aac atg ttc gag agc tac atc 491
 Gln Lys Thr Ala Arg Ser Asn Met Asp Asn Met Phe Glu Ser Tyr Ile

SEQUENCE LISTING.txt

130				135				140								
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ctg Leu 160	gag Glu	gcg Ala	gag Glu	ctt Leu	ggc Gly 165	aac Asn	atg Met	cag Gln	ggg Gly	ctg Leu 170	gtg Val	gag Glu	gac Asp	ttc Phe	aag Lys 175	587
aac Asn	aag Lys	tat Tyr	gag Glu	gat Asp 180	gag Glu	atc Ile	aat Asn	aag Lys	cgt Arg 185	aca Thr	gag Glu	atg Met	gag Glu	aac Asn 190	gaa Glu	635
ttt Phe	gtc Val	ctc Leu	atc Ile 195	aag Lys	aag Lys	gat Asp	gtg Val	gat Asp 200	gaa Glu	gct Ala	tac Tyr	atg Met	aac Asn 205	aag Lys	gta Val	683
gag Glu	ctg Leu	gag Glu 210	tct Ser	cgc Arg	ctg Leu	gaa Glu	ggg Gly 215	ctg Leu	acc Thr	gac Asp	gag Glu	atc Ile 220	aac Asn	ttc Phe	ctc Leu	731
agg Arg	cag Gln 225	cta Leu	tat Tyr	gaa Glu	gag Glu	gag Glu 230	atc Ile	cgg Arg	gag Glu	ctg Leu	cag Gln 235	tcc Ser	cag Gln	atc Ile	tcg Ser	779
gac Asp 240	aca Thr	tct Ser	gtg Val	gtg Val	ctg Leu 245	tcc Ser	atg Met	gac Asp	aac Asn	agc Ser 250	cgc Arg	tcc Ser	ctg Leu	gac Asp	atg Met 255	827
gac Asp	agc Ser	atc Ile	att Ile	gct Ala 260	gag Glu	gtc Val	aag Lys	gca Ala	cag Gln 265	tac Tyr	gag Glu	gat Asp	att Ile	gcc Ala 270	aac Asn	875
cgc Arg	agc Ser	cgg Arg	gct Ala 275	gag Glu	gct Ala	gag Glu	agc Ser	atg Met 280	tac Tyr	cag Gln	atc Ile	aag Lys	tat Tyr 285	gag Glu	gag Glu	923
ctg Leu	cag Gln	agc Ser 290	ctg Leu	gct Ala	ggg Gly	aag Lys	cac His 295	ggg Gly	gat Asp	gac Asp	ctg Leu	cgg Arg 300	cgc Arg	aca Thr	aag Lys	971
act Thr 305	gag Glu	atc Ile	tct Ser	gag Glu	atg Met	aac Asn 310	cgg Arg	aac Asn	atc Ile	agc Ser	cgg Arg 315	ctc Leu	cag Gln	gct Ala	gag Glu	1019
att Ile 320	gag Glu	ggc Gly	ctc Leu	aaa Lys	ggc Gly 325	cag Gln	agg Arg	gct Ala	tcc Ser	ctg Leu 330	gag Glu	gcc Ala	gcc Ala	att Ile	gca Ala 335	1067
gat Asp	gcc Ala	gag Glu	cag Gln	cgt Arg 340	gga Gly	gag Glu	ctg Leu	gcc Ala	att Ile 345	aag Lys	gat Asp	gcc Ala	aac Asn	gcc Ala 350	aag Lys	1115
ttg Leu	tcc Ser	gag Glu	ctg Leu 355	gag Glu	gcc Ala	gcc Ala	ctg Leu	cag Gln 360	cgg Arg	gcc Ala	aag Lys	cag Gln	gac Asp 365	atg Met	gcg Ala	1163
cgg Arg	cag Gln	ctg Leu 370	cgt Arg	gag Glu	tac Tyr	cag Gln	gag Glu 375	ctg Leu	atg Met	aac Asn	gtc Val	aag Lys 380	ctg Leu	gcc Ala	ctg Leu	1211
gac atc	gag atc	gcc acc	tac agg	aag ctg	ctg ctg	gag ggc	gag gag	gag gag	agc							1259

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Asp	Ile	Glu	Ile	Ala	Thr	Tyr	Arg	Lys	Leu	Leu	Glu	Gly	Glu	Glu	Ser		
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Arg	Leu	Glu	Ser	Gly	Met	Gln	Asn	Met	Ser	Ile	His	Thr	Lys	Thr	Thr		
400				405					410					415			
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Ser	Gly	Tyr	Ala	Gly	Gly	Leu	Ser	Ser	Ala	Tyr	Gly	Gly	Leu	Thr	Ser		
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ccc	ggc	ctc	agc	tac	agc	ctg	ggc	tcc	agc	ttt	ggc	tct	ggc	gcg	ggc		1403
Pro	Gly	Leu	Ser	Tyr	Ser	Leu	Gly	Ser	Ser	Phe	Gly	Ser	Gly	Ala	Gly		
			435					440					445				
tcc	agc	tcc	ttc	agc	cgc	acc	agc	tcc	tcc	agg	gcc	gtg	gtt	gtg	aag		1451
Ser	Ser	Ser	Phe	Ser	Arg	Thr	Ser	Ser	Ser	Arg	Ala	Val	Val	Val	Lys		
			450				455					460					
aag	atc	gag	aca	cgt	gat	ggg	aag	ctg	gtg	tct	gag	tcc	tct	gac	gtc		1499
Lys	Ile	Glu	Thr	Arg	Asp	Gly	Lys	Leu	Val	Ser	Glu	Ser	Ser	Asp	Val		
465					470						475						
ctg	ccc	aag	tga	acagctgcgg	cagcccctcc	cagcctaccc	ctcctgcgct										1551
Leu	Pro	Lys	*														
480																	
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tgaggctcag	ccctagccct	cagcccacct	ggggagttaa	ctacctgggg	accccccttg												1671
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 <213> H. sapiens

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 35 40 45
 Leu Gly Gly Gly Tyr Gly Gly Ala Ser Gly Met Gly Gly Ile Thr Ala
 50 55 60
 Val Thr Val Asn Gln Ser Leu Leu Ser Pro Leu Val Leu Glu Val Asp
 65 70 75 80
 Pro Asn Ile Gln Ala Val Arg Thr Gln Glu Lys Glu Gln Ile Lys Thr
 85 90 95
 Leu Asn Asn Lys Phe Ala Ser Phe Ile Asp Lys Val Arg Phe Leu Glu
 100 105 110
 Gln Gln Asn Lys Met Leu Glu Thr Lys Trp Ser Leu Leu Gln Gln Gln
 115 120 125
 Lys Thr Ala Arg Ser Asn Met Asp Asn Met Phe Glu Ser Tyr Ile Asn
 130 135 140
 Asn Leu Arg Arg Gln Leu Glu Thr Leu Gly Gln Glu Lys Leu Lys Leu
 145 150 155 160
 Glu Ala Glu Leu Gly Asn Met Gln Gly Leu Val Glu Asp Phe Lys Asn
 165 170 175
 Lys Tyr Glu Asp Glu Ile Asn Lys Arg Thr Glu Met Glu Asn Glu Phe
 180 185 190
 Val Leu Ile Lys Lys Asp Val Asp Glu Ala Tyr Met Asn Lys Val Glu

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195      200      205
Leu Glu Ser Arg Leu Glu Gly Leu Thr Asp Glu Ile Asn Phe Leu Arg
210      215      220
Gln Leu Tyr Glu Glu Glu Ile Arg Glu Leu Gln Ser Gln Ile Ser Asp
225      230      235
Thr Ser Val Val Leu Ser Met Asp Asn Ser Arg Ser Leu Asp Met Asp
245      250      255
Ser Ile Ile Ala Glu Val Lys Ala Gln Tyr Glu Asp Ile Ala Asn Arg
260      265      270
Ser Arg Ala Glu Ala Glu Ser Met Tyr Gln Ile Lys Tyr Glu Glu Leu
275      280      285
Gln Ser Leu Ala Gly Lys His Gly Asp Asp Leu Arg Arg Thr Lys Thr
290      295      300
Glu Ile Ser Glu Met Asn Arg Asn Ile Ser Arg Leu Gln Ala Glu Ile
305      310      315
Glu Gly Leu Lys Gly Gln Arg Ala Ser Leu Glu Ala Ala Ile Ala Asp
325      330      335
Ala Glu Gln Arg Gly Glu Leu Ala Ile Lys Asp Ala Asn Ala Lys Leu
340      345      350
Ser Glu Leu Glu Ala Ala Leu Gln Arg Ala Lys Gln Asp Met Ala Arg
355      360      365
Gln Leu Arg Glu Tyr Gln Glu Leu Met Asn Val Lys Leu Ala Leu Asp
370      375      380
Ile Glu Ile Ala Thr Tyr Arg Lys Leu Leu Glu Gly Glu Glu Ser Arg
385      390      395
Leu Glu Ser Gly Met Gln Asn Met Ser Ile His Thr Lys Thr Thr Ser
405      410      415
Gly Tyr Ala Gly Gly Leu Ser Ser Ala Tyr Gly Gly Leu Thr Ser Pro
420      425      430
Gly Leu Ser Tyr Ser Leu Gly Ser Ser Phe Gly Ser Gly Ala Gly Ser
435      440      445
Ser Ser Phe Ser Arg Thr Ser Ser Ser Arg Ala Val Val Val Lys Lys
450      455      460
Ile Glu Thr Arg Asp Gly Lys Leu Val Ser Glu Ser Ser Asp Val Leu
465      470      475
Pro Lys

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 <213> H. sapiens

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26

<210> 6
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<210> 7
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SEQUENCE LISTING.txt

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26

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<211> 7

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Ala Glu Gln Xaa Gly Glu Leu
1 5

<210> 9

<211> 14

<212> PRT

<213> H. sapiens

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Ile Ala Asp Ala Glu Gln Arg Gly Glu Leu Ala Ile Lys Asp
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